Report on the genetic constitution of human chromosome 19

H. Mohrenweiser and K. Johnson

Two hundred seventy genes (240 cloned) and 150 ESTs have been mapped to human chromosome 19. Five hundred forty STSs, (160 ESTs, 230 PCR-based polymorphic markers and 150 PCR-based non-polymorphic sites) have been assigned to chromosome 19. The average spacing between STSs is 110kb.

Over 290 polymorphic markers have been assigned to chromosome 19. 230 markers are PCR-based and 165 have heterozygosities >0.50; the distance between markers with heterozygosities of >0.5 is 365kb. Four independent genetic maps are available - Weber et al. (Am. J. Hum. Genet. 53:1079, 1993; CHLC (Hum Mol Genet 4:1837, 1995), Genethon (Cytogenet Cell Genet 71:158, 1995) and Utah Marker Development Group (Am J Hum Genet 57:619, 1995). Only a few markers are common to any pair of maps.

A metric physical map, based on assembly of cosmids into contigs where order of and distance between contigs is known, has been constructed by Lawrence Livermore National Laboratory (Nature Genetics 11:422, 1995). The backbone of this map is a set of 245 cosmids, FISH mapped using decondensed sperm pronuclear chromatin as the hybridization target, so that both the order of and distance between cosmids is known (Genomics 30:187, 1995). Ordered cosmid contigs spanning 37Mb (>80% of the chromosome) are in EcoRI restriction maps. The order of 150 of the genetic markers is known from localization on the physical map.

Disease genes continue to be the focus of significant attention on chromosome 19. Mutations in the COMP gene were identified in both PSACH and EDM1 patients. The localizations of MHP1 and CADASIL were refined while <EA2>, <DFNA4> and OFC3 gene were assigned to chromosome 19. DM was revisited with the cloning of <DMAHP>, a homeodomain protein associated with the unstable triplet repeat element of DM.

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